



INTERNATIONAL WORKSHOP on
**FAMILIAL ADULT
MYOCLONIC EPILEPSY**

NAPLES ITALY
ROYAL CONTINENTAL HOTEL
27TH-28TH MAY 2022



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Awarded to Antonietta Coppola

ENDORSEMENT





Dear colleagues and friends, we are very glad to welcome you to the International Workshop on Familial Adult Myoclonic Epilepsy (FAME/BAFME)

We wish you a great meeting and a delightful time in Naples
Antonietta and Pasquale

COURSE DIRECTORS



Antonietta Coppola



Pasquale Striano

FACULTY

Betul Baykan *Istanbul*
Mark Bennett *Melbourne*
Samuel Berkovic *Melbourne*
Leonilda Bilo *Naples*
Francesco Brancati *Rome*
Laura Canafoglia *Milan*
Mar Carreno *Barcelona*
Gianpiero Cavalleri *Dublin*
Sirio Cocozza *Naples*
Antonietta Coppola *Naples*
Mark Corbett *Adelaide*
Christel Depienne *Essen*
Raffaele Dubbioso *Naples*
Silvana Franceschetti *Milan*
Beatriz Gonzalez Giraldez *Madrid*
Edouard Hirsch *Strasbourg*
Akio Ikeda *Kyoto*
Karl Martin Klein *Calgary*
Laura Licchetta *Bologna*
Fiore Manganelli *Naples*
Roberto Michelucci *Bologna*
Christopher Pearson *Toronto*
Ingrid Scheffer *Melbourne*
Josè Serratosa *Madrid*
Isabel Silveira *Porto*
Pasquale Striano *Genoa*
Antonio Suppa *Rome*
Marina Tijssen *Groningen*
Shoji Tsuji *Tokyo*
Arn van den Maagdenberg *Leiden*
Anne-Fleur van Rootselaar *Amsterdam*
Liana Veneziano *Rome*
Federico Zara *Genoa*

INTERNATIONAL WORKSHOP
on FAMILIAL ADULT MYOCLONIC EPILEPSY



FRIDAY 27TH MAY

09.00 Registration of the participants

10.00 Welcome and introduction to the Workshop:
Antonietta Coppola
Leonilda Bilo
Pasquale Striano
Fiore Manganelli

FIRST SESSION:
Myoclonic epilepsies

Chairs: **Samuel Berkovic, Pasquale Striano**

10.30 Overview on myoclonic epilepsies
Ingrid Scheffer

11.00 Classification, clinical and neurophysiological
features, prognosis
Silvana Franceschetti

11.30 Drug treatment of myoclonus
Mar Carreno

12.00 Lunch

SECOND SESSION:

Electroclinical features and history of FAME/BAFME

Chairs: **Ingrid Scheffer, Antonio Suppa**

13.30 From BAFME to FAME: history of the disease across the world
Samuel Berkovic

13.50 Clinical findings, disease course and comorbidities
Akio Ikeda

14.10 Neurophysiological findings
Marina Tijssen

14.30 Discussion

15.00 Coffee break

THIRD SESSION:

Diagnostic investigations

Chairs: **Josè Serratosa, Karl Martin Klein**

15.30 Genetics of FAME: the history
Shoji Tsuji

15.50 Genetics of FAME: the state of the art
Mark Corbett

16.10 Neuropathology and Neuroimaging
Anne-Fleur van Rootselaar

16.30 Treatment options
Edouard Hirsch

16.50 Animal and cellular studies in (myoclonic) epilepsies
Arn van den Maagdenberg

17.10 Discussion

17.30 **Round table: Patients meet the Experts**

SATURDAY 28TH MAY

FOURTH SESSION:

FAME and Beyond: Pentanucleotide Related Disorders

Chairs: **Christel Depienne, Federico Zara**

09.00 Repeat expansion disorders: an overview
Christopher Pearson

09.20 Emerging pentanucleotide related disorder: the SCA37 model
Isabel Silveira

09.40 Bioinformatic analysis for repeat expansion
in short read sequencing data
Mark Bennett

10.00 Coffee break

10.30 **Research projects**

Chairs: **Gianpiero Cavalleri, Roberto Michelucci**

FAME: structural and functional implications
of aberrant sensory - motor excitability
Raffaele Dubbioso, Sirio Coccozza

Perampanel as adjunctive treatment in FAME 2 patients
Antonietta Coppola

Epidemiology of FAME in Italy. Unusual pentanucleotide repeat
expansions: disease or polymorphism?
Laura Licchetta, Liana Veneziano, Francesco Brancati

FAME 3 repeat Expansion structure revealed
by complementary long - read technologies
Christel Depienne

SAMD12 repeat expansion in a non - asian family
with negative repeat - primed PCR
Karl Martin Klein

Comparing progression and phenotypic severity
in FAME vs. PME
Laura Canafoglia

Long range PCR and targeted adaptive sampling
of FAME loci with nanopore sequencing
Mark Corbett

Previous misdiagnoses of patients with FAME
Betul Baykan

Refining the phenotype in FAME 3:
novel seizure types and clues to minimize misdiagnosis
Beatriz Gonzalez Giraldez

13.00 Closing remarks

13.30 Light lunch



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General Information

Congress venue
HOTEL ROYAL CONTINENTAL NAPOLI
VIA PARTENOPE, 38/44
80121 Naples
Meeting Room Mirabilis - First floor

Organizing Secretariat



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